Class 12: Genome Informatics

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Section 1. Proportion of G/G in a population

Downloaded a CSV File from Ensemble < https://useast.ensembl.org/Homo_sapiens/Variation/Sample?db=core;r=17:39894595-39895595;v=rs8067378;vdb=variation;vf=959672880;sample=MXL#373531_tablePanel <

Here we read the CSV file we obtained:

```
mxl <- read.csv("373531-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
head(mxl)
```

```
##   Sample..Male.Female.Unknown. Genotype..forward.strand. Population.s. Father
## 1                  NA19648 (F)                       A|A ALL, AMR, MXL      -
## 2                  NA19649 (M)                       G|G ALL, AMR, MXL      -
## 3                  NA19651 (F)                       A|A ALL, AMR, MXL      -
## 4                  NA19652 (M)                       G|G ALL, AMR, MXL      -
## 5                  NA19654 (F)                       G|G ALL, AMR, MXL      -
## 6                  NA19655 (M)                       A|G ALL, AMR, MXL      -
##   Mother
## 1      -
## 2      -
## 3      -
## 4      -
## 5      -
## 6      -
```

```
table(mxl$Genotype..forward.strand.)
```

```
##
## A|A A|G G|A G|G
##  22  21  12   9
```

```
table(mxl$Genotype..forward.strand.) / nrow(mxl) * 100
```

```
##
##     A|A     A|G     G|A     G|G
## 34.3750 32.8125 18.7500 14.0625
```

Now let's look at a different population, GBR, "great Britian."

```
gbr <- read.csv("373522-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
```

Find portion of GIG

```
round(table(gbr$Genotype..forward.strand.) / nrow(gbr) *100,2)
```

```
##
```

```
##   A|A   A|G   G|A   G|G
## 25.27 18.68 26.37 29.67
```

This varient associated with childhood asthma is more prevelant in the GBR population than the MXL population.

Let's dig deeper into this!

Section 4: Population Scale Analysis

One sample is obviously not enough to know what is happening in a population. You are interested in assessing genetic differences on a population scale.

> Q13. How many samples do we have? What is their median expression levels for each of these geneotypes?

```r
expr <- read.table("rs8067378_ENSG00000172057.6.txt")
head(expr)
```

```
##    sample geno      exp
## 1 HG00367  A/G 28.96038
## 2 NA20768  A/G 20.24449
## 3 HG00361  A/A 31.32628
## 4 HG00135  A/A 34.11169
## 5 NA18870  G/G 18.25141
## 6 NA11993  A/A 32.89721
```

```r
nrow(expr)
```

```
## [1] 462
```

Sample size per genotype:

```r
table(expr$geno)
```

```
##
## A/A A/G G/G
## 108 233 121
```

Median Expression Levels for each genotype:

```r
library(dplyr)
```

```
##
## Attaching package: 'dplyr'

## The following objects are masked from 'package:stats':
##
##     filter, lag

## The following objects are masked from 'package:base':
##
##     intersect, setdiff, setequal, union
```

```r
summary_expr <- expr %>%
  group_by(geno) %>%
  summarise(
    median_expression = median(exp, na.rm=TRUE)
  )
```

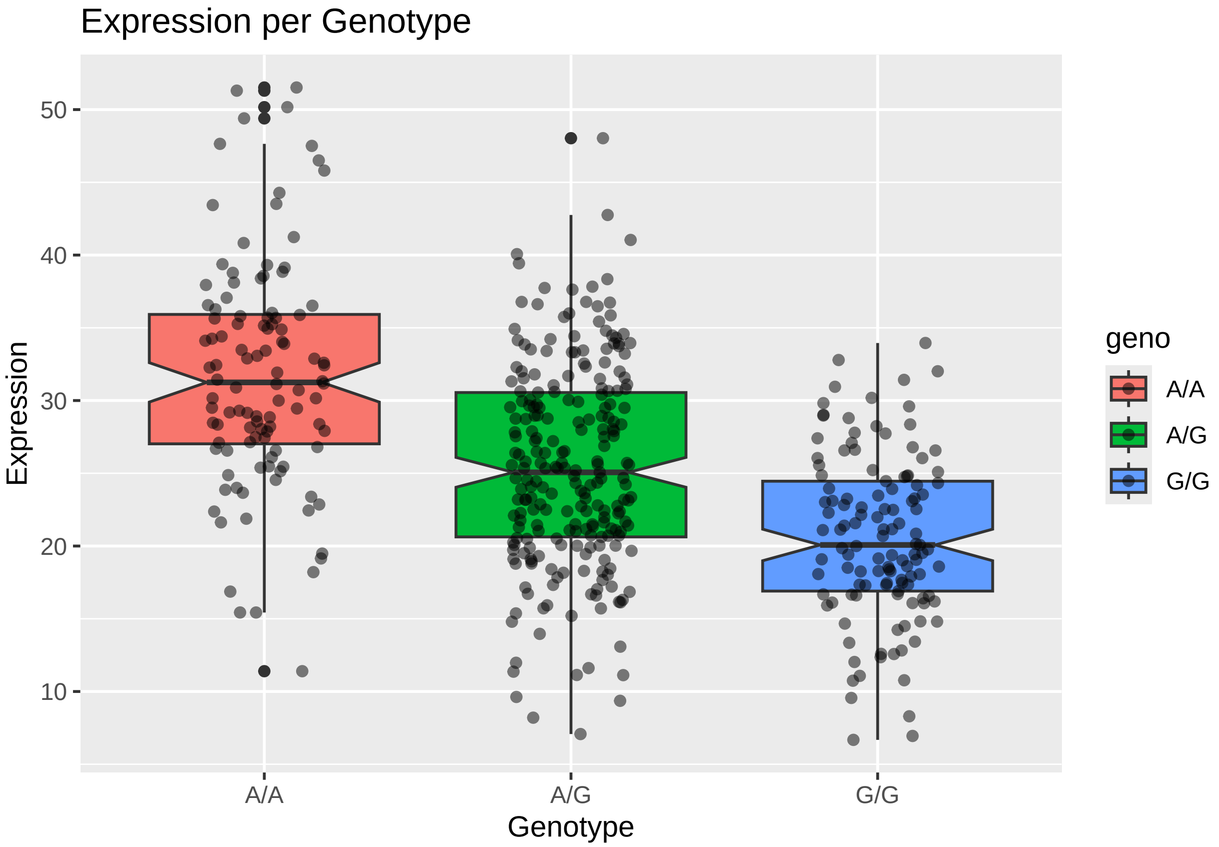
```r
print(summary_expr)
```

```
## # A tibble: 3 x 2
##   geno  median_expression
##   <chr>             <dbl>
## 1 A/A                31.2
## 2 A/G                25.1
## 3 G/G                20.1
```

Q14. Generate a boxplot with a box per genotype.

```r
library(ggplot2)

ggplot(expr) + aes(geno, exp, fill=geno) +
  geom_boxplot(notch=TRUE) +
  geom_jitter(alpha=0.5, width=0.2) +
  labs(title="Expression per Genotype",x="Genotype", y="Expression")
```



Q14 continued. What could you infer from the relative expression value between A/A and G/G displayed in this plot? Does the SNP effect the expression of ORMDL3?

A|A has a higher median expression than G|G displayed in the plot. Yes, the SNP does effect the expression of ORMDL3, with the A allese suggesting higher association with ORMDL3.